What is supplemental newborn screening?

The supplemental newborn screening study is a research project that identifies several metabolic disorders. These metabolic disorders are not currently a part of the mandatory routine screening that every baby receives while at the hospital.

Participation in the supplemental newborn screening study is voluntary. In the hospital, you will be asked to sign a consent form to let the hospital staff know whether or not you want your baby to participate. Agreeing to participate means that the blood sample already taken for the mandatory routine screening will be tested for more disorders. If you participate, there is no additional cost to you or your insurance company, and it does not require taking any more blood from your baby.

What are metabolic disorders?

Metabolic disorders affect the body's ability to produce or break down compounds such as proteins, fats, or carbohydrates into smaller substances the body needs for energy, growth, and repair. Too much of certain substances or too little of others can cause major health problems. If identified early, some of these conditions can be treated before they cause serious health problems. Treatment may include the close monitoring of your baby's health, medication, dietary supplements, or special diets.

Metabolic disorders have varying degrees of severity. Some of the conditions identified by the study may not cause significant health problems or require treatment, others, as of yet, have no truly effective treatment.

Will I find out my baby's results? Probably not.

During the study, only unusual results will be reported to your baby's pediatrician. Unusual results indicate that a disorder is suspected or additional testing is needed.

What happens if a disorder is suspected or results are unusual?

Your baby's doctor will be contacted if there are any unusual results. Further evaluation will be needed to find out if your newborn has a disorder. Your baby will be referred to a doctor who specializes in the treatment of metabolic disorders. The specialist will examine your baby and possibly order more tests. If your newborn has a disorder, the specialist will assist you and your pediatrician in treating your baby's special health care needs.

What are the benefits of participating in the supplemental newborn screening study (also known as the tandem mass spectrometry research study)?

- Supplemental screening may identify a metabolic disorder that is not included in the mandatory routine screening program.
- Early identification of a metabolic disorder may prevent death as well as physical and mental disability through effective treatment.
- Supplemental screening is provided at no additional cost during the study period.
- If further testing is required to identify a metabolic disorder in your baby, this diagnostic testing is usually covered by your health insurance or Medicaid.
- By participating in the study, you are helping the Utah Newborn Screening Program determine which disorders to add to the mandatory routine screening program in the future. This screening will benefit newborns with these disorders as well as their families.

What are the risks of participating in the supplemental newborn screening study?

- Treatment services are not covered by the Utah Newborn Screening Program. These services are usually covered by your health insurance or Medicaid.
- Approximately three out of every 1,000 newborns will have an unusual result. Most babies with unusual tandem mass spectrometry results (MS/MS) will not have a metabolic disorder.
- A small number of newborns with a metabolic disorder may not be identified by the MS/MS screening study. For these babies, treatment could be delayed until after symptoms appear. Be sure to tell your doctor if your baby does not appear well or seems to be developing more slowly than expected.
- If your baby's blood sample is unsuitable for testing due to contamination, improper temperature submission, or other technical issues, supplemental (MS/MS) screening will not occur. A new blood sample will be collected by the hospital and then only mandatory routine screening will occur. Your baby's doctor will be notified of any specimen that is unsuitable for testing and can arrange a retest for supplemental (MS/MS) screening at your request.

What if someone in your family has a metabolic disorder?

Please discuss this issue with your pediatrician. If there is a family history of a metabolic condition or a special concern, you should obtain supplemental testing outside of, or in addition to, the research study. Your doctor can obtain more information on available testing laboratories, diagnosis, and the treatment of metabolic disorders by contacting the Metabolic Clinic at the University of Utah. Optional supplemental screening is also offered for a fee at the Mayo Clinic Medical Center in Rochester, Minnesota and at Neo Gen Screening in Bridgeville, Pennsylvania.

Disorders Potentially Detected

All of the disorders below are **autosomal recessive disorders**, which means that, even though neither parent is usually affected, each parent must pass an affected gene to their baby for the disorder to appear. There is a one-in-four chance, or a 25% chance, that this will happen each time the couple conceives. Metabolic disorders affect the body's ability to produce or break down compounds such as amino acids, organic acids, and fatty acids into smaller substances the body needs for energy, growth, and repair.

Amino Acid Disorders

The terms "amino acidemia" and "amino aciduria" refer to disorders in amino acid metabolism (the break down process that provides energy or heat for body functions). Amino acids are the chemical building blocks of human proteins. Proteins are responsible for cell functions in the body. In order for amino acids to work, specific enzymes must be present. Aminoacidurias are disorders resulting from a lack of the proper enzymes needed for amino acid metabolism and transport. This deficiency results in abnormally large quantities of amino acids in the urine or blood, which can be toxic to the body.

Symptoms vary by disorder and may include slow development, vomiting, diarrhea, abnormal urine odor or color, and the build up of acid in the body (acidosis). Disorders in amino acid metabolism can result in mental retardation.

Treatments may include special diets, vitamins at high doses, replacement of the deficient enzyme, and medication. Prompt treatment may prevent serious problems from developing.

Disorders:

- Maple syrup urine disease (MSUD)
- Homocystinuria/cystathionine beta-synthetase deficiency
- Citrullinemia/argininosuccinic acid synthetase deficiency
- Argininosuccinyl-CoA lyase deficiency (ASAL)
- Phenylketonuria (PKU)
- Argininemia/arginase deficiency
- Tyrosinemia

Organic Acid Disorders

Organic acid disorders can be referred to as "organic acidemias" or "organic acidurias." Organic acids are a group of chemicals that are used in critical metabolic processes in the body. Organic acid disorders usually result from a missing or malfunctioning step in amino acid catabolism (chemical break down) due to a lack of enzyme activity.

Symptoms vary by disorder and may include poor feeding, vomiting, low blood sugar, drowsiness, seizures, brain disease, and coma.

Treatment may include a special diet or medication to remedy the problems caused by the deficient enzyme activity.

Disorders:

- Propionic acidemia
- Methylmalonic acidemia
- Isobutyryl-CoA dehydrogenase deficiency
- Isovaleric acidemia
- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMGCoA)
- Glutaric acidemia - type-1 (GA-1)
- 2-methylbutyryl-CoA dehydrogenase deficiency
- 3-methylcrotonyl-CoA carboxylase deficiency (3MCC)
- Beta-ketothiolase deficiency (BKD)

Fatty Acid Oxidation Disorders

Fatty acids are a component of fat in table food and the fat in our body's tissues. Oxidation is the process that breaks down fatty acids to release energy needed for body functions. Each step of the oxidation process is set in motion by a specific enzyme. Fatty acid oxidation disorders occur when one of these enzymes is missing.

Symptoms vary by disorder and may include drowsiness, poor skin tone, vomiting, low blood sugar, brain disease, liver failure, and muscle problems. Without treatment, these problems can lead to coma and even death.

Treatments include low-fat diets, avoiding fasting, and maintaining a regular intake of sugar, carnitine, and other dietary supplements.

Disorders:

- Short chain acyl-CoA dehydrogenase (SCAD) deficiency
- Medium chain acyl-CoA dehydrogenase (MCADD) deficiency
- Very long chain acyl-CoA dehydrogenase (VLCAD) deficiency
- 3-Hydroxy long chain acyl-CoA dehydrogenase (LCHAD) deficiency and trifunctional protein deficiency
- Carnitine palmitoyl transferase deficiency - type 2 (CPT-2)
- Carnitine-acylcarnitine translocase deficiency (CAT)
- Carnitine transporter deficiency
- Multiple acyl-CoA dehydrogenase deficiency (MADD)/ glutaric acidemia - type-2 (GA-2)
- Carnitine palmitoyl transferase deficiency - type 1 (CPT-1)

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